



Fig. 1

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Table 1

Number and Frequency (%) of *ACTN3* Genotypes and Frequency (%) of *ACTN3* Alleles in Controls and Elite Sprint/Power and Endurance Athletes

GROUP (n)	No. (%) WITH GENOTYPE			ALLEL FREQUENCY (%)	
	RR	RX	XX	R	X
<b>Male:</b>					
Controls (134)	40 (30)	73 (54)	21 (16)	57	43
Sprint (72)	38 (53)	28 (39)	6 (8)	72	28
Endurance (122)	34 (28)	63 (52)	25 (20)	54	46
<b>Female:</b>					
Controls (292)	88 (30)	147 (50)	57 (20)	55	45
Sprint (35)	15 (43)	20 (57)	0 (0)	71	29
Endurance (72)	26 (36)	25 (35)	21 (29)	53	47
<b>Total:</b>					
Controls (436)	130 (30)	226 (52)	80 (18)	56	44
Sprint (107)	53 (50)	48 (45)	6 (6)	72	28
Endurance (194)	60 (31)	88 (45)	46 (24)	54	46

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**TABLE 2: Genotyping of R577X in ACTN3 in Caucasians Elite Athletes.**

Strength	Sport	ID	Sport Institute	Total Number	577RR (%)	577RX (%)	577XX (%)
Endurance	Rower	RT492	AIS	64	22 (34.4%)	28 (43.8%)	14 (21.8%)
		RT556					
Endurance	Triathloner	RT977	AIS	13	3 (23.1%)	8 (61.5%)	2 (15.4%)
		RT989					
Endurance	Cyclist	RT990	AIS	9	4 (44.4%)	2 (22.2%)	3 (33.3%)
		RT998					
Endurance	Track Cyclist	KN246	AIS	22	7 (31.8%)	7 (31.8%)	8 (36.4%)
		KN275					
Endurance	Marathon	KN310	AIS	1	0	0	1
Endurance	All above		AIS	108	36 (33.3%)	45 (41.7%)	27 (25.0%)
Sprint	Swimmer	RT901	AIS	45	17 (37.8%)	25 (55.6%)	3 (6.6%)
		RT1018					
Sprint	Track Cyclist	KN246	AIS	8	4 (50.0%)	3 (37.5%)	1 (12.5%)
		KN275					
Sprint	Athletics	KN276	AIS	30	16 (53.3%)	13 (43.3%)	1 (3.3%)
		KN309					
Sprint	All above		AIS	83	37 (44.6%)	41 (49.4%)	5 (6.0%)
Africa Zulu				88	69 (78.4%)	18 (20.5%)	1 (1.1%)
Australian Caucasian Control				152	46 (30.0%)	78 (52.0%)	28 (18%)

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**Table 3 SNPs identified in the ACTN3 gene to date**  
**NCBI SNP CLUSTER ID**

rs2229456

rs2229455

rs2229454

rs2228325

rs1126675

rs7949754

rs7924602

rs5792393

rs4990284

rs4990283

rs4013815

rs3937320

rs3837428

rs3814736

rs3814735

rs3782080

rs2511217

rs2511216

rs2509559

rs2509558

rs2305537

rs2305534

rs2290463

rs2275998

rs2096583

rs2000939

rs1815739

rs1791690

rs1671064

rs1188610

rs679228

rs678397

rs677488

rs647476

rs647029

rs618838

rs607736

rs597626

rs544021

rs540874

rs538330

rs531490

rs509556

rs490998

rs13897

rs4576

rs1189338

rs1201433

rs640213

rs3737525

rs3178740

rs3180065

rs3180064

rs3180063

rs3867132

rs608504

rs610293

rs3825065

[www.ncbi.nlm.nih.gov/SNP/snp\\_ref](http://www.ncbi.nlm.nih.gov/SNP/snp_ref)

TSC: The SNP Consortium website

**TABLE 4. Symbols, full names, and cytogenic location of nuclear and mitochondrial genes of the 2002 Human Gene Map for Performance and Health-Related Fitness Phenotypes.**

**Gene or Locus Name Location**

**A B**

*ACADVL* Acyl coenzyme A dehydrogenase, very long chain 17p13-p11

*ACE* Angiotensin I converting enzyme 17q23

*ADRA2A* Alpha-2A-adrenergic receptor 10q24-q26

*ADRB1* Adrenergic, beta-1-, receptor 10q24-q26

*ADRB2* Beta-2-adrenergic receptor 5q31-q32

*ADRB3* Beta-3-adrenergic receptor 8p12-p11.2

*AGT* Angiotensinogen 1q42-q43

*ANG* Angiogenin, ribonuclease, RNase A family, 5 14q11.1-q11.2

*APOE* Apolipoprotein E 19q13.2

*ATP1A2* ATPase, Na<sub>+</sub>/K<sub>-</sub> transporting, alpha-2 polypeptide 1q21-q23

*ATP1B1* ATPase, Na<sub>+</sub>/K<sub>-</sub> transporting, beta 1 polypeptide 1q22-q25

*BDKRB2* Bradykinin receptor B2 14q32.1-q32.2

**C D E F G**

*CASQ2* Calsequestrin 2 (cardiac muscle) 1p13.3-p11

*CFTR* Cystic fibrosis transmembrane conductance regulator, ATP-binding cassette (sub-family C, member 7) 7q31.2

*CKM* Creatine kinase, muscle 19q13.2-q13.3

*CNTF* Ciliary neurotrophic factor 11q12.2

*CPT2* Carnitine palmitoyltransferase 2 1p32

*COL1A1* Collagen, type I, alpha 1 17q21.3-q22.1

*EDN1* Endothelin 1 6p24.1

*ENO3* Enolase 3, (beta, muscle) 17pter-p11

*FABP2* Fatty acid binding protein 2 4q28-q31

*FGA* Fibrinogen, A alpha polypeptide 4q28

*FGB* Fibrinogen, B beta polypeptide 4q28

*GDF8 (MSTN)* Growth differentiation factor 8 (myostatin) 2q32.2

*GNB3* Guanine nucleotide binding protein (G protein), beta polypeptide 3 12p13

**H I K L M**

*HLA-A* Major histocompatibility complex, class I, A 6p21.3

*HP* Haptoglobin 16q22.1

*IGF1* Insulin-like growth factor I 12q22-q23

*IGF2* Insulin-like growth factor 2 11p15.5

*IL-6 Interleukin-6*

*KCNQ1* K<sub>+</sub>voltage-gated channel, KQT-like subfamily, member 1 11p15.5

*LDHA* Lactate dehydrogenase A 11p15.4

*LPL* Lipoprotein lipase 8p22

*MTCO1* Cytochrome c oxidase I mtDNA 5904-7445

*MTCO3* Cytochrome c oxidase III mtDNA 9207-9990

*MTCYB* Cytochrome b mtDNA 14747-15887

*MTND1* NADH dehydrogenase 1 mtDNA 3307-4262

*MTND4* NADH dehydrogenase 4 mtDNA 10760-12137

*MTND5* NADH dehydrogenase 5 mtDNA 12337-14148  
*MTTE* Transfer RNA, mitochondrial, glutamic acid mtDNA 14674-14742  
*MTTI* Transfer RNA, mitochondrial, isoleucine mtDNA 4263-4331  
*MTTK* Transfer RNA, mitochondrial, lysine mtDNA 8295-8364  
*MTTL1* Transfer RNA, mitochondrial, leucine 1 (UUR) mtDNA 3230-3304  
*MTTL2* Transfer RNA, mitochondrial, leucine 2 (CUN) mtDNA 12266-12336  
*MTTM* Transfer RNA, mitochondrial, methionine mtDNA 4402-4469  
*MTTT* Transfer RNA, mitochondrial, threonine mtDNA 15888-15953  
*MTTY* Transfer RNA, mitochondrial, tyrosine mtDNA 5826-5891  
*MyHC myosin Heavy-chain*  
N O P Q R S T U V  
*NOS3* Nitric oxide synthase 3 (endothelial cell) 7q36  
*NPY* Neuropeptide Y 7p15.1  
*PAI1* Plasminogen activator inhibitor 1 7q21.3-q22  
*PFKM* Phosphofructokinase, muscle 12q13.3  
*PGAM2* Phosphoglycerate mutase 2 (muscle) 7p13-p12  
*PGK1* Phosphoglycerate kinase 1 Xq13  
*PHKA1* Phosphorylase kinase, alpha 1 (muscle) Xq12-q13  
*PON1* Paraoxonase 1 7q21.3  
*PPARA* Peroxisome proliferative activated receptor, alpha 22q13.31  
*PPARG* Peroxisome proliferative activated receptor, gamma 3p25  
*PYGM* Phosphorylase, glycogen, muscle 11q12-q13.2  
*RYR2* Ryanodine receptor 2 (cardiac) 1q42.1-q43  
*SGCA* Sarcoglycan, alpha (50kDa dystrophin-associated glycoprotein) 17q21  
*S100A1* S100 calcium binding protein A1 1q21  
*SUR* Sulfonylurea receptor 11p15.1  
*TGFB1* Transforming growth factor beta 1 19q13.2  
*UCP2* Uncoupling protein 2 11q13  
*UCP3* Uncoupling protein 3 11q13  
*VDR* Vitamin D (1,25-dihydroxyvitamin D3) receptor 12q12-q14  
The gene symbols, names and cytogenetic locations are from the Locus Link web site  
(<http://www.ncbi.nlm.nih.gov/LocusLink>) available from the National Center for  
Biotechnology  
Information (NCBI). For mitochondrial DNA, locations are from the human  
mitochondrial genome data base (<http://www.mitomap.org>).

TABLE 5. Endurance phenotypes and case-control studies (DNA polymorphisms).

Gene	Location	Athletes			Controls		
		N	Sports	Freq.	N	Freq.	P
<i>ADRA2A</i>	10q24-q26	140	Endurance	6.7/6.7: 0.77	141	6.7/6.7: 0.62	0.037
				6.7/6.3: 0.21		6.7/6.3: 0.34	
				6.3/6.3: 0.02		6.3/6.3: 0.04	
				6.7: 0.88		6.7: 0.8	0.011
				6.3: 0.12		6.3: 0.2	
<i>ACE</i>	17q23	64	Endurance	II: 0.30	118	II: 0.18	0.03
				ID: 0.55		ID: 0.51	
				DD: 0.16		DD: 0.32	
				I: 0.57		I: 0.43	0.02
				D: 0.43		D: 0.57	
	79	Running		I: 0.57	Ref. Pop. Ref.	I: 0.49	0.039
				D: 0.43		D: 0.51	
				NA		NA	
				II: 0.25		Pop. Ref.	0.003
	25	Mountain-eering			1248	II: 0.16	0.0009
				ID: 0.58		ID: 0.45	
				DD: 0.17		DD: 0.39	
				I: 0.54		I: 0.38	
				D: 0.46		D: 0.62	
	56	Elite swimmers (subsample of 103 swimmers)		II: 0.15	1248	II: 0.24	0.004
				ID: 0.39		ID: 0.49	
				DD: 0.46		DD: 0.27	
				I: 0.34		I: 0.48	
				D: 0.66		D: 0.52	

Reference: Perusse et al. 2003 "The human gene map for performance and health-related fitness phenotypes: the 2002 update" Med. Sci. Sports Exerc. 35: 1248-1264.

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TABLE 6. Genotype and allelic frequencies of *ACTN3* 577R/X alleles in human populations.

Ethnic group	No. of chromosomes	No. of genotypes		Relative allele frequency of 577X
		RX	XX	
Asian	56	14	7	0.5+0.07
Javanese	96	28	12	0.54+0.05
Native American	14	2	2	0.43+0.14
Asia/Americas	166	44	21	0.52+0.04
Hispanic	64	16	5	0.41+0.06
White	214	47	21	0.42+0.03
Europe	278	63	26	0.41+0.03
Aboriginal	174	33	9	0.29+0.03
Australian				
PNG Highlander	78	16	6	0.36+0.05
Australasia	252	49	15	0.31+0.03
African American	90	12	6	0.27+0.05
African Bantu	156	14	1	0.10+0.05
Africa	246	56	7	0.16+0.05
Unknown	152	50	11	0.47
Total	1094	232	80	